young children and infants and their practical significance therefore needs to be emphasized in the teaching of pediatrics so that pediatricians can interact meaningfully with pathologists in this somewhat difficult area of bone marrow interpretation.

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Megalocornea–Mental Retardation Syndrome

A 42-month-old girl presented with global developmental delay and an abnormally large right eye. She was the product of a consanguineous marriage. She had a history suggestive of hypotonia since infancy with delayed developmental milestones. Examination revealed microcephaly and short stature. The right eye had megalocornea (corneal diameter >13 mm) and a normal iris. The left eye was normal. She had preaxial polydactyly of the right hand. Apart from hypotonia, the rest of the neurological and systemic examination was normal. Development assessment in the individual domains by the Early Developmental Profile were 19 months in gross motor, 23 months in fine motor, 21 months in cognition, 9.8 months in language, 24 months in social/emotional and 16.3 months in feeding. The development quotient was 44.9 with a disproportionate delay in language.

On investigation, her bone age was normal and there was no skeletal abnormality except the polydactyly. BERA revealed bilateral sensorineural deafness, Investigations revealed no other associated abnormalities. A diagnosis of Megalocornea-Mental retardation syndrome (type 5) was made.

Megalocornea-Mental Retardation syndrome (MMR) was first reported by Neuhaus, et al.(1). It is an extremely rare disorder with autosomal recessive inheritance. Most cases appear sporadically. The syndrome is characterized by distinctive ocular abnormalities,
congenital hypotonia, and varying degrees of mental retardation. In some cases, additional abnormalities may also be present. The range and severity of symptoms and physical findings vary and have raised questions regarding the nosology of the syndrome and the issue of heterogeneity versus variability(2).

Most infants have bilateral megalocornea. Additional ocular abnormalities may exist including iris hypoplasia and iridodonesis. Other abnormalities occasionally include short stature, seizures, and craniofacial malformations (frontal bossing, hypertelorism, a long upper lip, hypoplastic mandible, and large ears).

Clinical classification of MMR includes: type 1 (Neuhauser) with iris hypoplasia and minor anomalies; type 2 (Frank-Temtamy) with camptodactyly, scoliosis and growth retardation; type 3 (Verloes) with normal irides, severe hypotonia, macrocephaly and minor anomalies; type 4 (Frydman) with normal irides, megalencephaly and obesity; and type 5, unclassifiable cases(3). Other synonyms for MMR are megalocornea developmental retardation dysmorphic syndrome, Neuhauser syndrome and megalocornea macrocephaly mental and motor retardation syndrome(4).

To our knowledge this is the first case to have been reported from India. The small number of published cases may be due to the fact that the association of these two signs has often escaped attention until now(5).

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